



SEQUENCE LISTING

<110> MELKI, Judith
MUNNICH, Arnold

<120> Spinal Muscular Atrophy Diagnostic Methods

<130> 2121-0140P

<140> 09/109,082

<141> 1998-07-02

<150> 08/545,196

<151> 1995-10-19

<160> 65

<170> PatentIn version 3.2

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<211> 347

<212> DNA

<213> Homo sapiens

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<213> Homo sapiens

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aaacaatcaa tattaaagaa ttttgatgcc aaaactatta gataaaaggt taatctacat 180
ccctactaga attctcatac ttaactgggt gggtatgtgg aagaaacata ctttcacaat 240
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tttattgtga tatgggataa cctaggcata ctgcactgta cactctgaca tatgaagtgc 360
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ccactgcaag aaaaccttaa ctgcagccta ataattgttt tctttgggat aactttttaa 180
gtacattaaa agactatcaa cttaatttct gatcataatt tgttgaataa aataagtaaa 240
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<213> Homo sapiens

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aaacaatcaa tattaagaa ttttgatgcc aaaactatta gataaaaggт таатctacat 180
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<223> R111 primer/probe characteristic of exon 8 of the T-BCD541 gene.

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<210> 6
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 <212> DNA
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 <223> 541C770 primer/probe characteristic of exon 8 of the T-BCD541 gene.

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Ile Trp Asp Asp Thr Ala Leu Ile Lys Ala Tyr Asp Lys Ala Val Ala
 35 40 45

Ser Phe Lys His Ala Leu Lys Asn Gly Asp Ile Cys Glu Thr Ser Gly
 50 55 60

Lys Pro Lys Thr Thr Pro Lys Arg Lys Pro Ala Lys Lys Asn Lys Ser
 65 70 75 80

Gln Lys Lys Asn Thr Ala Ala Ser Leu Gln Gln Trp Lys Val Gly Asp
 85 90 95

Lys Cys Ser Ala Ile Trp Ser Glu Asp Gly Cys Ile Tyr Pro Ala Thr
 100 105 110

Ile Ala Ser Ile Asp Phe Lys Arg Glu Thr Cys Val Val Val Tyr Thr
 115 120 125

Gly Tyr Gly Asn Arg Glu Glu Gln Asn Leu Ser Asp Leu Leu Ser Pro
 130 135 140

Ile Cys Glu Val Ala Asn Asn Ile Glu Gln Asn Ala Gln Glu Asn Glu
 145 150 155 160

Asn Glu Ser Gln Val Ser Thr Asp Glu Ser Glu Asn Ser Arg Ser Pro
 165 170 175

Gly Asn Lys Ser Asp Asn Ile Lys Pro Lys Ser Ala Pro Trp Asn Ser
 180 185 190

Phe Leu Pro Pro Pro Pro Pro Met Pro Gly Pro Arg Leu Gly Pro Gly
 195 200 205

Lys Pro Gly Leu Lys Phe Asn Gly Pro Pro Pro Pro Pro Pro Pro
 210 215 220

Pro Pro His Leu Leu Ser Cys Trp Leu Pro Pro Phe Pro Ser Gly Pro
 225 230 235 240

Pro Ile Ile Pro Pro Pro Pro Pro Ile Cys Pro Asp Ser Leu Asp Asp
 245 250 255

Ala Asp Ala Leu Gly Ser Met Leu Ile Ser Trp Tyr Met Ser Gly Tyr
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 gattctgaca tttgggatga tacagcactg ataaaagcat atgataaagc tgtggcttca 180
 ttttaagcatg ctctaaagaa tgggtgacatt tgtgaaactt cgggtaaacc aaaaaccaca 240
 cctaaaagaa aacctgctaa gaagaataaa agccaaaaga agaatactgc agcttcctta 300
 caacagtgga aagttgggga caaatgttct gccatttggg cagaagacgg ttgcatttac 360
 ccagctacca ttgcttcaat tgattttaag agagaaacct gtgttgtggg ttacactgga 420
 tatggaaata gagaggagca aaatctgtcc gatctacttt cccaatctg tgaagtagct 480
 aataatatag aacagaatgc tcaagagaat gaaaatgaaa gccaaagtttc aacagatgaa 540
 agtgagaact ccagggtctcc tggaaataaa tcagataaca tcaagcccaa atctgctcca 600
 tggaaccctt ttctccctcc accacccccc atgccagggc caagactggg accaggaaag 660
 ccagggtctaa aattcaatgg cccaccaccg ccaccgccac caccaccacc ccacttacta 720
 tcatgctggc tgcctccatt tccttctgga ccaccaataa ttccccacc acctccata 780
 tgtccagatt ctcttgatga tgctgatgct ttgggaagta tgttaatttc atggtacatg 840
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 agaaacgatc agacagatct ggaatgtgaa gcgttataga agataactgg cctcatttct 1020
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tatcttctat atgttttaaaa gtatataata aaaatatatta attttttttt aaaaaaaaaa	1500
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aaaaaaaaaa aaaaaaaaaa aa	1582

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ccactgcaag aaaaccttaa ctgcagccta ataattgttt tctttgggat aacttttaaa	180
gtacattaaa agactatcaa ctttaatttct gatcatattt tgttgaataa aataagtaaa	240
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gttaaaaagt tcagatgtta gaaagttgaa aggttaatgt aaaacaatca atattaaaga	540
atTTTgatgc caaaactatt agataaaaagg ttaatctaca tccctactag aattctcata	600
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 <212> DNA
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gattctgaca tttgggatga tacagcactg ataaaagcat atgataaagc tgtggcttca	180
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aaaaaaaaaa aaaaaaaaaa aaaaaaaaaa aaaaaaaaaa aaaaaaaaaa aaaaaaaaaa	1560
aaaaaaaaaa aaaaaaaaaa aa	1582

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 <213> Homo sapiens

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 gaatctaact ggtggacatg gctgttcatt gtactgtttt tttctatctt ctatatgttt 1380
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<210> 14
 <211> 372
 <212> DNA
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<220>
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 <222> (14)..(14)
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 <223> n is a, c, g, or t

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 <222> (84)..(84)
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 <223> n is a, c, g, or t

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 <222> (319)..(319)
 <223> n is a, c, g, or t

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 <222> (332)..(332)
 <223> n is a, c, g, or t

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 <222> (348)..(348)
 <223> n is a, c, g, or t

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 gggtgataag agtgggaccc tgtntcaaaa catacacaca cacacacaca cacacacaca 120
 cacacacaca cacactctct ctctctctct ctctctctct ctctctctct ctctctctca 180
 aaaacacttg gtctgttatt tttncgaaat tgtcagtcac agttatctgt tagaccaaag 240
 ctgngtaagn acatttatta cattgcctcc tacaacttca tcagctaata tatttgctat 300
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 ctgaggaaag ga 372

<210> 15
 <211> 294
 <212> DNA
 <213> Artificial Sequence

<220>
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tccccctccc ccttggaaaa gtaagtaagc tctactagg aatttaaaac ctgcttgatc 240
tatataaaga caaacaagga aagacaaaca tgggggagcagg aaggaaggca gatc 294

<210> 16
<211> 141
<212> DNA
<213> Artificial Sequence

<220>
<223> C171 marker nucleotide sequence.

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cacacacaga cttaatctgt ttacagaaat aaaaggaata aaataccgtt tctactatac 120
acaaaaacta gccatcttga c 141

<210> 17
<211> 305
<212> DNA
<213> Artificial Sequence

<220>
<223> AFM157xd10 marker nucleotide sequence.

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tgtgtgtgtg tgtgtatgtt tgcttgact gtaaaaacaa ttgcaacatc aacagaaata	120
aaaattaaag gaataattct cctccgactc tgccgttcca tccagtgaaa ctcttcattc	180
tggggtaaag ttccttcagt tctttcatag ataggtatat acttcataag tcaaacaatc	240
aggctgggtg cagtagctca tgcctgtaat cccagccctt tgggaggccg agctgggcag	300
atcga	305

<210> 18
 <211> 350
 <212> DNA
 <213> Artificial Sequence

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 <223> C161 marker nucleotide sequence.

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 <222> (69)..(69)
 <223> n is a, c, g, or t

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 <223> n is a, c, g, or t

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 <223> n is a, c, g, or t

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<220>
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 <223> n is a, c, g, or t

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<222> (298)..(298)
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 ggcatactgt gtattgggat ggggttagaa caggtgttct acccaagaca tttacttaaa 180
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 aaagcatgaa agtatttatg cttgattttt tttttttnact catagcttca tagtgganca 300
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<210> 19
 <211> 278
 <212> PRT
 <213> Homo sapiens

<400> 19
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 20 25 30

Ile Trp Asp Asp Thr Ala Leu Ile Lys Ala Tyr Asp Lys Ala Val Ala
 35 40 45

Ser Phe Lys His Ala Leu Lys Asn Gly Asp Ile Cys Glu Thr Ser Gly
 50 55 60

Lys Pro Lys Thr Thr Pro Lys Arg Lys Pro Ala Lys Lys Asn Lys Ser
 65 70 75 80

Gln Lys Lys Asn Thr Ala Ala Ser Leu Gln Gln Trp Lys Val Gly Asp
 85 90 95

Lys Cys Ser Ala Ile Trp Ser Glu Asp Gly Cys Ile Tyr Pro Ala Thr
 100 105 110

Ile Ala Ser Ile Asp Phe Lys Arg Glu Thr Cys Val Val Val Tyr Thr
 115 120 125

Gly Tyr Gly Asn Arg Glu Glu Gln Asn Leu Ser Asp Leu Leu Ser Pro

130	135	140
Ile Cys Glu Val Ala Asn Asn Ile Glu Gln Asn Ala Gln Glu Asn Glu		
145	150	155 160
Asn Glu Ser Gln Val Ser Thr Asp Glu Ser Glu Asn Ser Arg Ser Pro		
	165	170 175
Gly Asn Lys Ser Asp Asn Ile Lys Pro Lys Ser Ala Pro Trp Asn Ser		
	180	185 190
Phe Leu Pro Pro Pro Pro Pro Met Pro Gly Pro Arg Leu Gly Pro Gly		
	195	200 205
Lys Pro Gly Leu Lys Phe Asn Gly Pro Pro Pro Pro Pro Pro Pro Pro		
	210	215 220
Pro Pro His Leu Leu Ser Cys Trp Leu Pro Pro Phe Pro Ser Gly Pro		
225	230	235 240
Pro Ile Ile Pro Pro Pro Pro Pro Ile Cys Pro Asp Ser Leu Asp Asp		
	245	250 255
Ala Asp Ala Leu Gly Ser Met Leu Ile Ser Trp Tyr Met Ser Gly Tyr		
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His Thr Gly Tyr Tyr Met		
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Gln Glu Asp Thr Val Leu Phe Arg Arg Gly Thr Gly Gln Ser Asp Asp	
15 20 25	

tct gac att tgg gat gat aca gca ttg ata aaa gct tat gat aaa gct	146
Ser Asp Ile Trp Asp Asp Thr Ala Leu Ile Lys Ala Tyr Asp Lys Ala	
30 35 40	
gtg gct tcc ttt aag cat gct cta aag aac ggt gac att tgt gaa act	194
Val Ala Ser Phe Lys His Ala Leu Lys Asn Gly Asp Ile Cys Glu Thr	
45 50 55	
cca gat aag cca aaa ggc aca gcc aga aga aaa cct gcc aag aag aat	242
Pro Asp Lys Pro Lys Gly Thr Ala Arg Arg Lys Pro Ala Lys Lys Asn	
60 65 70 75	
aaa agc caa aag aag aat gcc aca act ccc ttg aaa cag tgg aaa gtt	290
Lys Ser Gln Lys Lys Asn Ala Thr Thr Pro Leu Lys Gln Trp Lys Val	
80 85 90	
ggt gac aag tgt tct gct gtt tgg tca gaa gac ggc tgc att tac cca	338
Gly Asp Lys Cys Ser Ala Val Trp Ser Glu Asp Gly Cys Ile Tyr Pro	
95 100 105	
gct act att acg tcc att gac ttt aag aga gaa acc tgt gtc gtg gtt	386
Ala Thr Ile Thr Ser Ile Asp Phe Lys Arg Glu Thr Cys Val Val Val	
110 115 120	
tat act gga tat gga aac aga gag gag caa aac tta tct gac cta ctt	434
Tyr Thr Gly Tyr Gly Asn Arg Glu Glu Gln Asn Leu Ser Asp Leu Leu	
125 130 135	
tcc ccg acc tgt gaa gta gct aat agt aca gaa cag aac act cag gag	482
Ser Pro Thr Cys Glu Val Ala Asn Ser Thr Glu Gln Asn Thr Gln Glu	
140 145 150 155	
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Asn Glu Ser Gln Val Ser Thr Asp Asp Ser Glu His Ser Ser Arg Ser	
160 165 170	
ctc aga agt aaa gca cac agc aag tcc aaa gct gct ccg tgg acc tca	578
Leu Arg Ser Lys Ala His Ser Lys Ser Lys Ala Ala Pro Trp Thr Ser	
175 180 185	
ttt ctt cct cca cca ccc cca atg cca ggg tca gga tta gga cca gga	626
Phe Leu Pro Pro Pro Pro Pro Met Pro Gly Ser Gly Leu Gly Pro Gly	
190 195 200	
aag cca ggt cta aaa ttc aac ggc ccg ccg ccg ccg cct cca cta ccc	674
Lys Pro Gly Leu Lys Phe Asn Gly Pro Pro Pro Pro Pro Pro Leu Pro	
205 210 215	
cct ccc ccc ttc ctg ccg tgc tgg atg ccc ccg ttc cct tca gga cca	722
Pro Pro Pro Phe Leu Pro Cys Trp Met Pro Pro Phe Pro Ser Gly Pro	
220 225 230 235	
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Pro Ile Ile Pro Pro Pro Pro Pro Ile Ser Pro Asp Cys Leu Asp Asp	
240 245 250	

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Thr Asp Ala Leu Gly Ser Met Leu Ile Ser Trp Tyr Met Ser Gly Tyr	
255 260 265	

cac act ggc tac tat atg ggt ttc aga caa aat aaa aaa gaa gga aag	866
His Thr Gly Tyr Tyr Met Gly Phe Arg Gln Asn Lys Lys Glu Gly Lys	
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Cys Ser His Thr Asn	
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Asp Thr Ala Leu Ile Lys Ala Tyr Asp Lys Ala Val Ala Ser Phe Lys
35 40 45

His Ala Leu Lys Asn Gly Asp Ile Cys Glu Thr Pro Asp Lys Pro Lys
50 55 60

Gly Thr Ala Arg Arg Lys Pro Ala Lys Lys Asn Lys Ser Gln Lys Lys
65 70 75 80

Asn Ala Thr Thr Pro Leu Lys Gln Trp Lys Val Gly Asp Lys Cys Ser
85 90 95

Ala Val Trp Ser Glu Asp Gly Cys Ile Tyr Pro Ala Thr Ile Thr Ser
100 105 110

Ile Asp Phe Lys Arg Glu Thr Cys Val Val Val Tyr Thr Gly Tyr Gly
115 120 125

Asn Arg Glu Glu Gln Asn Leu Ser Asp Leu Leu Ser Pro Thr Cys Glu
130 135 140

Val Ala Asn Ser Thr Glu Gln Asn Thr Gln Glu Asn Glu Ser Gln Val

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Ser Thr Asp Asp	Ser Glu His Ser Ser Arg Ser Leu Arg Ser Lys Ala					
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His Ser Lys Ser Lys Ala Ala Pro Trp Thr Ser Phe Leu Pro Pro Pro						
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Pro Pro Met Pro Gly Ser Gly Leu Gly Pro Gly Lys Pro Gly Leu Lys						
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Phe Asn Gly Pro Pro Pro Pro Pro Pro Leu Pro Pro Pro Pro Phe Leu						
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Pro Cys Trp Met Pro Pro Phe Pro Ser Gly Pro Pro Ile Ile Pro Pro						
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Pro Pro Pro Ile Ser Pro Asp Cys Leu Asp Asp Thr Asp Ala Leu Gly						
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Ser Met Leu Ile Ser Trp Tyr Met Ser Gly Tyr His Thr Gly Tyr Tyr						
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Met Ala Met Ser
1

agc ggc ggc agt ggt ggc ggc gtc ccg gag cag gag gat tcc gtg ctg 163
Ser Gly Gly Ser Gly Gly Gly Val Pro Glu Gln Glu Asp Ser Val Leu
5 10 15 20

ttc cgg cgc ggc aca ggc cag gtgaggtcgc agccagtgca gtctccctat 214
Phe Arg Arg Gly Thr Gly Gln
25

tagcgtcttc agcacccttc ttccggccca actctccttc cgcagtgtaa ttttggtatg 274

tgtggattaa gatgactctt ggtactaaca tacattttct gattaaacct atctgnacat 334

gagttgtttt tatttcttac cttttccag agc gat gat tct gac att tgg gat 387
Ser Asp Asp Ser Asp Ile Trp Asp
30 35

gat aca gca ctg ata aaa gca tat gat aaa gct gtg gct tca ttt aag 435
Asp Thr Ala Leu Ile Lys Ala Tyr Asp Lys Ala Val Ala Ser Phe Lys
40 45 50

gtatgaaatg cttgnttagt cgttttctta ttttctcggtt attcatttgg aaaggaattg 495

ataacatacg ataaagtgtt aaaggtgctt tctgaggtga cggagccttg agactagctt 555

atagtagtaa ctgggttatg tctgtacttt tattctgtgc accaccctgt aacatgtaca	615
tttttattcc tattttcgta g cat gct cta aag aat ggt gac att tgt gaa	666
His Ala Leu Lys Asn Gly Asp Ile Cys Glu	
55 60	
act tcg ggt aaa cca aaa acc aca cct aaa aga aaa cct gct aag aag	714
Thr Ser Gly Lys Pro Lys Thr Thr Pro Lys Arg Lys Pro Ala Lys Lys	
65 70 75	
aat aaa agc caa aag aag aat act gca gct tcc tta caa cag	756
Asn Lys Ser Gln Lys Lys Asn Thr Ala Ala Ser Leu Gln Gln	
80 85 90	
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caaaagaaat gtgtgcatgt atatatcttt gatttctttt gtag tgg aaa gtt ggg	932
Trp Lys Val Gly	
95	
gac aaa tgt tct gcc att tgg tca gaa gac ggt tgc att tac cca gct	980
Asp Lys Cys Ser Ala Ile Trp Ser Glu Asp Gly Cys Ile Tyr Pro Ala	
100 105 110	
acc att gct tca att gat ttt aag aga gaa acc tgt gtt gtg gtt tac	1028
Thr Ile Ala Ser Ile Asp Phe Lys Arg Glu Thr Cys Val Val Val Tyr	
115 120 125	
act gga tat gga aat aga gag gag caa aat ctg tcc gat cta ctt tcc	1076
Thr Gly Tyr Gly Asn Arg Glu Glu Gln Asn Leu Ser Asp Leu Leu Ser	
130 135 140	
cca atc tgt gaa gta gct aat aat ata gaa cag aat gct caa gag	1121
Pro Ile Cys Glu Val Ala Asn Asn Ile Glu Gln Asn Ala Gln Glu	
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ctgcatatatt tttcttttta aag aat gaa aat gaa agc caa gtt tca aca gat	1294
Asn Glu Asn Glu Ser Gln Val Ser Thr Asp	
160 165	
gaa agt gag aac tcc agg tct cct gga aat aaa tca gat aac atc aag	1342
Glu Ser Glu Asn Ser Arg Ser Pro Gly Asn Lys Ser Asp Asn Ile Lys	
170 175 180	
ccc aaa tct gct cca tgg aac tct ttt ctc cct cca cca ccc ccc atg	1390
Pro Lys Ser Ala Pro Trp Asn Ser Phe Leu Pro Pro Pro Pro Met	
185 190 195 200	
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Pro Gly Pro Arg Leu Gly Pro Gly Lys	
205	

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ccacaaatat tctgggtaat tattttttatc cttttgggtt tgagtccttt ttattcctat	1557
catattgaaa ttggtaagtt aatttttcctt tgaaatatc cttatag cca ggt cta	1613
	Pro Gly Leu
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aaa ttc aat ggc cca cca ccg cca ccg cca cca cca cca ccc cac tta	1661
Lys Phe Asn Gly Pro Pro Pro Pro Pro Pro Pro Pro Pro Pro Pro His Leu	
215 220 225	
cta tca tgc tgg ctg cct cca ttt cct tct gga cca cca gtaagtaaaa	1710
Leu Ser Cys Trp Leu Pro Pro Phe Pro Ser Gly Pro Pro	
230 235 240	
aagagtatag gttagatttt gctttcacat acaatttgat aattaccaga ctttactttt	1770
tgtttactgg atataaacia tatctttttc tgtctccag ata att ccc cca cca	1824
	Ile Ile Pro Pro Pro
	245
cct ccc ata tgt cca gat tct ctt gat gat gct gat gct ttg gga agt	1872
Pro Pro Ile Cys Pro Asp Ser Leu Asp Asp Ala Asp Ala Leu Gly Ser	
250 255 260	
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Met Leu Ile Ser Trp Tyr Met Ser Gly Tyr His Thr Gly Tyr Tyr Met	
265 270 275	
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gtaaatttat aaaatactac ttgaactgca gcctaataat tgttttcttt gggataactt	2040
ttaaagtaca ttaaaagact atcaacttaa tttctgatca tattttgttg aataaaataa	2100
gtaaaatgtc ttgtgaaaca aaatgctttt taacatccat ataaagctat ctatatatag	2160
ctatctatgt ctatatagct atttttttta acttctttt attttctta cag ggt	2216
	Gly
ttc aga caa aat caa aaa gaa gga agg tgc tca cat tcc tta aat	2261
Phe Arg Gln Asn Gln Lys Glu Gly Arg Cys Ser His Ser Leu Asn	
280 285 290	
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tggtgaaaac aaatgttttt gaacagttaa aaagttcaga tggtaaaaag ttgaaagggt	2381
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cacaataaag agcttttaga tatgatgcca ttttatatca ctagtaggca gaccagcaga	2561

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aagcctctgg ttctaatttc tcatttgcag gaaatgctgg catagagcag cactaaatga	2741
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atatctacac acacacacac acacacacca cactggagtt cgagacgagg cctaagcaac	180
atgccgaaac cccgtctcta ctaaatacaa aaaatagctg agcttggtgg cgcacgccta	240
tagtcctagc tactggggag gctgaggtgg gaggatcgct tgagcccaag aagtcgaggc	300
tgcagtgagc cgagatcgcg ccgctgcact ccagcctgag cgacagggcg aggctctgtc	360
tcaaaacaaa caaacaaaaa aaaaaaggaa aggaaatata acacagtga atgaaaggat	420
tgagagaaat gaaaaatata cagccacaa atgtgggagg gcgataacca ctcgtagaaa	480
gcgtgagaag ttactacaag cggctctccc gggcacgta ctgttccgct cccagaagcc	540
ccgggcgcgg gaagtcgtca ctcttaagaa gggacggggc cccacgctgc gcacccgcgg	600
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 1 5 10 15

Ile Tyr Pro Ala Thr Ile Ala Ser Ile Asp Phe Lys Arg Glu Thr Cys
 20 25 30

Val Val Val Tyr Thr Gly Tyr Gly Asn Arg Glu Glu Gln Asn Leu Ser
 35 40 45

Asp Leu Leu Ser Pro Ile Cys Glu Val Ala Asn Asn Ile Glu Gln Asn
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Ala Gln Glu
 65

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Asn Glu Asn Glu Ser Gln Val Ser Thr Asp Glu Ser Glu Asn Ser Arg
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Ser Pro Gly Asn Lys Ser Asp Asn Ile Lys Pro Lys Ser Ala Pro Trp
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Asn Ser Phe Leu Pro Pro Pro Pro Pro Met Pro Gly Pro Arg Leu Gly
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Pro Gly Lys

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20 25 30

Thr Gly Tyr Tyr Met
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